

IN THE CLAIMS

1-4. (Canceled)

5. (Withdrawn) A therapeutic agent for Rothmund-Thomson syndrome, which contains as the effective ingredient a DNA encoding RecQ4 helicase.

6. (Withdrawn) A therapeutic agent for Rothmund-Thomson syndrome, which contains as the effective ingredient a RecQ4 helicase

7. (Previously presented) A diagnostic agent for Rothmund-Thomson syndrome, wherein the agent comprises a preparation of antibodies which specifically bind to RecQ4 helicase as shown in SEQ ID NO: 4.

8. (Withdrawn) A method for the diagnosis of Rothmund-Thomson syndrome, characterized by detecting mutations in the DNA encoding RecQ4 helicase or the expression regulatory region thereof.

9. (Withdrawn) The method for the diagnosis of Rothmund-Thomson syndrome *in* claim 8, comprising the steps of:

(a) preparing DNA samples from patients;

(b) amplifying the prepared DNA samples using the DNA of claim 4 as a primer and determining the base sequence; and

(c) comparing the determined base sequence with that of a healthy normal person.

10. (Withdrawn) The method for the diagnosis of Rothmund-Thomson syndrome in claim 8, comprising the steps of:

(a) preparing RNA samples from patients;

(b) separating the prepared RNA samples according to their size;

(c) using the DNA of claim 4 as a probe, hybridizing it to

separated RNAs; and

(d) detecting the hybridized RNA and comparing the results with that of a normal, healthy person.

11. (Withdrawn) The method for the diagnosis of Rothmund-Thomson syndrome in claim 8, comprising the steps of:

- (a) preparing DNA samples from patients;
- (b) amplifying the prepared DNA samples using the DNA of claim 4 as a primer;
- (c) dissociating the amplified DNA into single-stranded DNA;
- (d) fractionating the dissociated single-stranded DNAs on a non-denaturing gel; and
- (e) comparing the mobility of the fractionated single-stranded DNA on the gel with that of a healthy normal person.

12. (Withdrawn) The method for the diagnosis of Rothmund-Thomson syndrome in claim 8, comprising the steps of:

- (a) preparing DNA samples from the patient;
- (b) amplifying the prepared DNA samples using oligonucleotides comprising a base that forms a base pair with the mutated base specific to Rothmund-Thomson syndrome in the DNA encoding RecQ4 helicase, or the expression regulatory region thereof, as at least one of the primers; . and
- (c) detecting the amplified DNA fragment.

13. (Withdrawn) The method for the diagnosis of Rothmund-Thomson syndrome in claim 8, comprising the steps of:

- (a) preparing DNA samples from patients;
- (b) amplifying the prepared DNA samples using a pair of DNA of claim 4 which is prepared so as to flank the mutated base specific to Rothmund-Thomson syndrome as the primer;
- (c) hybridizing to the amplified product a pair of oligonucleotides selected from the group of:
 - (i) an oligonucleotide synthesized such that the base forming a base pair with the mutated base in the amplified product corresponds to the 3' -terminus, and an oligonucleotide synthesized such that the neighboring (on the 3'side) base to said 3'-terminus corresponds to the 5'-terminus;
 - (ii) an oligonucleotide synthesized such that the base forming a base pair with the base of a normal healthy person which corresponds to the mutated base in the amplified product corresponds to the 3'-terminus, and an oligonucleotide synthesized such that the neighboring (on the 3'side) base to said 3'-terminus corresponds to the 5'-terminus;
 - (iii) an oligonucleotide synthesized such that the base forming a base pair with the mutated base in the amplification product corresponds to the 5'-terminus, and an oligonucleotide synthesized such that the neighboring (on the 5' site) base to said 5'-terminus corresponds to the 3'-terminus: and
 - (iv) an oligonucleotide synthesized such that the base forming

a base pair with the base of a normal healthy person which corresponds to the mutated base in the amplified product corresponds to the 5' -terminus, and an oligonucleotide synthesized such that the neighboring (on the 5' site} base to said 5'-terminus corresponds to the 3'-terminus;

(d) ligating the oligonucleotides; and

(e) detecting the ligated oligonucleotide.

14. (Withdrawn) The method for the diagnosis of Rothmund-Thomson syndrome in claim 8, comprising the steps of:

(a) preparing protein samples from patients;

(b) contacting an antibody against RecQ4 helicase with the prepared protein sample; and

(c) detecting proteins binding to said antibody.

15. (Previously presented) The diagnostic agent of claim 7 wherein the antibody binds to amino acids 907 to 1208 of SEQ ID NO: 4.